Cleveland Clinic

Somatic & Germline Mutations

Germline mutations are changes to your DNA that you inherit from the egg and sperm cells during conception. Somatic mutations are changes to your DNA that happen after conception to cells other than the egg and sperm. Mutations can lead to genetic conditions that affect your health.

Overview

What are DNA mutations?

A mutation is a change to your DNA sequence, which is the information your cells receive to be able to perform properly.

Changes to your DNA happen when your cells divide and replicate. Most changes to a person's DNA don't affect their genetic makeup and won't lead to any health problems, but some mutations cause genetic conditions that could affect your health.

There are thousands of possible genetic mutations that could occur when your cells divide and replicate. Two types of genetic mutations include:

- Germline mutations.
- Somatic mutations.

What are germline mutations?

Germline mutations occur in a parent's reproductive cells (egg or sperm). These mutations change the genetic material that the child receives from their parent (hereditary). You can inherit germline mutations from either parent.

What are somatic mutations?

Somatic mutations are a change to a person's DNA that occurs after conception to any cell that isn't a germ cell (egg or sperm cell). Somatic mutations don't pass from parents to their children (not hereditary) and happen sporadically or randomly, without the mutation existing in a person's family history. They also can't pass to future generations.

Function

How do somatic and germline mutations affect my body?

Most mutations don’t cause problems for us, but some mutations create symptoms of disease. Genetic conditions are disorders that are caused by changes to your genome. Your genome is made up of your DNA, genes and chromosomes.

Can I inherit a mutation?

You can inherit germline genetic mutations. In this type of mutation, the DNA change happens in reproductive cells (egg or sperm). The sperm and egg pass from parents to their children, so the mutation is inherited.

You can’t inherit somatic mutations because they occur randomly in cells that aren't the sperm or egg.

Anatomy

Where is DNA in my body?

DNA exists in every cell in your body (there are trillions) and holds your genetic code. Your genetic code is your body’s instruction manual.

What does DNA look like?

DNA is a structure made up of four bases:

- Adenine (A).
- Cytosine (C).
- Thymine (T).

- Guanine (G).

The bases form pairs (base pairs): A with T and C with G. The base pairs connect with a sugar molecule and a phosphate molecule (to form a nucleotide). As the nucleotides form, they make a shape that looks like a spiral staircase (double helix) in your cells. The base pairs are the steps and the sugar and phosphate molecules are the handrails.

Where do germline mutations happen?

Germline mutations change the DNA in the reproductive cells. The egg and sperm combine at fertilization, and the cells copy themselves to create new cells that make an embryo. Parents who are carriers of a mutation can pass the mutation onto their children.

Where do somatic mutations happen?

The change to a person's DNA during a somatic mutation happens after fertilization in any cell of their body that isn't a sperm or egg cell (germ cells). Cells continuously copy and replace themselves in humans. If a mutation occurs, all cells that form from that affected cell will have that mutation in its DNA.

Conditions and Disorders

What are the common conditions and disorders caused by germline mutations?

Inherited conditions are the result of germline mutations. There are hundreds of them, but some common germline mutation conditions include:

- Sickle cell disease.
- Cystic fibrosis.

- Tay-Sachs disease.
- Huntington's disease.

What are the common conditions and disorders caused by somatic mutations?

Somatic mutations can cause conditions that can affect a person's health. Common somatic mutation conditions include:

- Skin cancer.
- Lung cancer.
- McCune-Albright syndrome.
- Sturge-Weber syndrome.

Is there a test to detect mutations?

Genetic tests detect mutations, which are changes to your genes, chromosomes or proteins. Genetic testing may identify what gene or chromosome specifically has a mutation. Genetic tests can help parents understand their risk of having a child with a genetic condition if there's a history of genetic conditions in their family.

Care

How do I prevent mutations?

You can take steps to reduce your risk of somatic mutations by:

- Wearing sunscreen when outdoors and reducing your exposure to UV rays.

- Using personal protective equipment like a mask and gloves when handling chemicals.
- Not smoking.
- Eating a healthy diet and exercising regularly.

You can’t prevent inherited germline mutations. To understand your risk of having a child with a germline mutation, talk to your healthcare provider about genetic testing.

A note from Cleveland Clinic

Most changes to your DNA don’t lead to genetic conditions, but some DNA changes could affect your health. If you want to understand your risk of having a child with a genetic condition, talk to your healthcare provider about genetic testing for germline mutations, especially if there’s a history of genetic conditions in your family. Take steps to prevent somatic mutations that aren't hereditary by wearing sunscreen, limiting UV exposure, avoiding dangerous chemicals and maintaining a healthy lifestyle.

Care at Cleveland Clinic

Do certain health conditions seem to run in your family? Are you ready to find out if you’re at risk? Cleveland Clinic’s genetics team can help.